



IDUA gene

iduronidase, alpha-L-

Normal Function

The *IDUA* gene provides instructions for producing an enzyme called alpha-L-iduronidase, which is essential for the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Through a process called hydrolysis, alpha-L-iduronidase uses water molecules to break down a molecule known as unsulfated alpha-L-iduronic acid, which is present in two GAGs called heparan sulfate and dermatan sulfate. Alpha-L-iduronidase is located in lysosomes, compartments within cells that digest and recycle different types of molecules.

Health Conditions Related to Genetic Changes

mucopolysaccharidosis type I

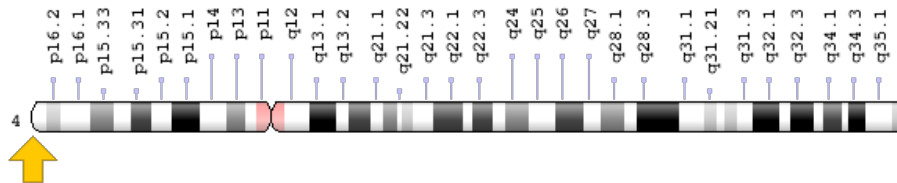
More than 100 mutations in the *IDUA* gene have been found to cause mucopolysaccharidosis type I (MPS I). Mutations that change one DNA building block (nucleotide) are the most common. All mutations that cause MPS I reduce or completely eliminate the function of alpha-L-iduronidase. It usually cannot be determined whether a certain mutation will cause severe or attenuated MPS I; however, people who do not produce any alpha-L-iduronidase have the severe form of this disorder.

The lack of alpha-L-iduronidase enzyme activity leads to the accumulation of heparan sulfate and dermatan sulfate within the lysosomes. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in MPS I. Researchers believe that the accumulated GAGs may also interfere with the functions of other proteins inside the lysosomes and disrupt the movement of molecules inside the cell.

Chromosomal Location

Cytogenetic Location: 4p16.3, which is the short (p) arm of chromosome 4 at position 16.3

Molecular Location: base pairs 986,997 to 1,004,557 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha-L-iduronidase
- IDUA_HUMAN

Additional Information & Resources

Educational Resources

- Eureka Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462>

GeneReviews

- Mucopolysaccharidosis Type I
<https://www.ncbi.nlm.nih.gov/books/NBK1162>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28IDUA%5BTIAB%5D%29+OR+%28alpha-L-iduronidase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- ALPHA-L-IDURONIDASE
<http://omim.org/entry/252800>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=IDUA%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5391
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3425>
- UniProt
<http://www.uniprot.org/uniprot/P35475>

Sources for This Summary

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Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/IDUA>

Reviewed: December 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services